SBDS gene

SBDS, ribosome assembly guanine nucleotide exchange factor

Normal Function

The *SBDS* gene provides instructions for making a protein whose function is unknown. Because mutations in this gene cause health problems affecting many body systems, researchers believe that the SBDS protein has an essential function in cells throughout the body.

Studies suggest that the SBDS protein may play a role in processing RNA, a molecule that is a chemical cousin of DNA. This protein may also be involved in building ribosomes, which are cellular structures that use the instructions encoded by RNA to create proteins. More research is needed to clarify the protein's role in these processes.

Health Conditions Related to Genetic Changes

Shwachman-Diamond syndrome

At least 20 mutations in the *SBDS* gene have been identified in people with Shwachman-Diamond syndrome. Most of these mutations result from an exchange of genetic material between the *SBDS* gene and a very similar, but nonfunctional, piece of DNA called a pseudogene, which is located very close to the *SBDS* gene on chromosome 7. This type of DNA exchange is called a gene conversion. The genetic material from the pseudogene contains errors that, when introduced into the *SBDS* gene, disrupt the way the gene's instructions are used to make a protein.

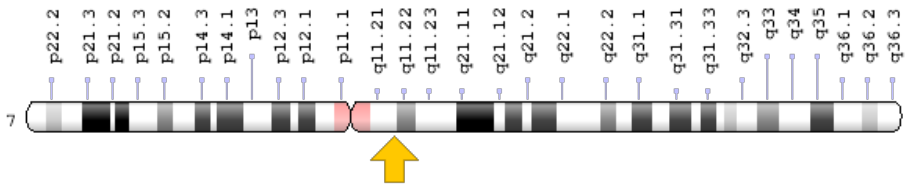
The two most common mutations in people with Shwachman-Diamond syndrome result from exchanges between the *SBDS* gene and the nearby pseudogene. One of these mutations, written as 258+2T>C, changes a single DNA building block (nucleotide) in a region of the gene known as intron 2. This mutation, which is called a splice-site mutation, prevents the production of any functional SBDS protein. The other common mutation, written as 183-184TA>CT, changes two nucleotides in the *SBDS* gene. This genetic change introduces a premature stop signal in the instructions for making the SBDS protein. It is unclear whether this mutation results in an abnormally shortened protein or prevents any protein from being made.

The features of Shwachman-Diamond syndrome result when mutations impair the normal function of the SBDS protein. Because the protein's function is unknown, researchers have not determined how these mutations underlie the bone marrow abnormalities, increased cancer risk, and other signs and symptoms of this condition.

Chromosomal Location

Cytogenetic Location: 7q11.21, which is the long (q) arm of chromosome 7 at position 11.21

Molecular Location: base pairs 66,987,703 to 66,995,601 on chromosome 7 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CGI-97
- FLJ10917
- SBDS ribosome assembly guanine nucleotide exchange factor
- SBDS_HUMAN
- Sdol1
- SDS
- Shwachman-Bodian-Diamond syndrome
- SWDS
- YLR022c

Additional Information & Resources

GeneReviews

- Shwachman-Diamond Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1756

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SBDS%5BTIAB%5D%29+OR+%28Shwachman-Bodian-Diamond+syndrome%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- SBDS GENE
  http://omim.org/entry/607444

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SBDS.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SBDS%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19440
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/51119
- UniProt
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